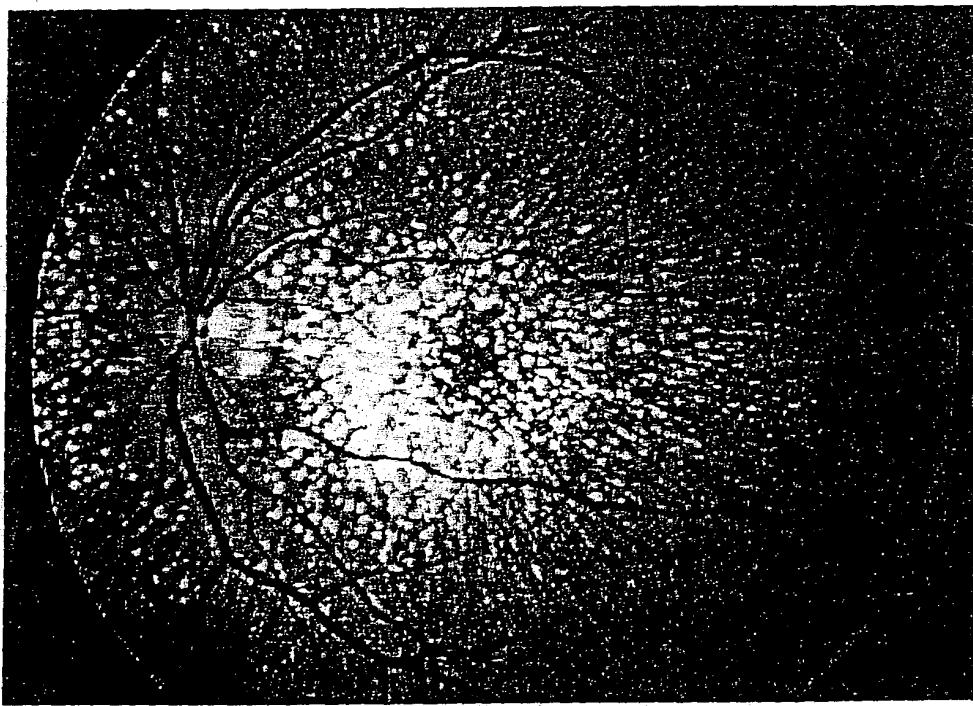


**SHEET 1 OF 12**

**Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS**

**Inventors: Stone et al. Ref. No. 21087.0019U3**



**FIG. 1**

SHEET 2 OF 12  
Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS  
Inventors: Stone *et al.* Ref. No. 21087.0019U3

p-tel  
D2S119  
D2S2227  
D2S2316  
SGC-31133  
WI-11560  
D38521  
stSG10198  
D2S2251  
SGC-35022  
D2S2352  
BCD1993  
SGC-34889  
WI-22280  
RP-S27A1  
WI-1332  
A009M26  
SGC-32447  
D2S2153  
WI-15186  
WI-16857  
133O18AAAT  
B-FODRIN 5'  
B-FODRIN 3'  
293J12CA  
133O18CA  
FBNL INTRO 2  
FBNL EXON 7  
FBNL EXON 9  
FBNL EXON 10  
FBNL INTRON 11  
322A4-33  
322A4 AAAT  
WI-6613  
322A4-29  
WI-6704  
SGC-34452  
WI-6241  
WI-18292  
stSG-42480  
D2S378  
SGC-31346  
WI-11399  
D2S1364  
SGC-34988  
D2S357  
D2S147  
cen

14cM ML RECOMBINATION INTERVAL, HEON, *et al.*, 1996  
5 cM DHHD RECOMBINATION INTERVAL, GREGORY *et al.*, 1996  
6.2 cM ML RECOMBINATION INTERVAL, EDWARDS *et al.*, 1998

INITIAL SWISS HAPLOTYPIC INTERVAL

AUSTRALIAN RECOMBINATION INTERVAL

FINAL SWISS HAPLOTYPIC INTERVAL

FIG. 2A

## SHEET 3 OF 12

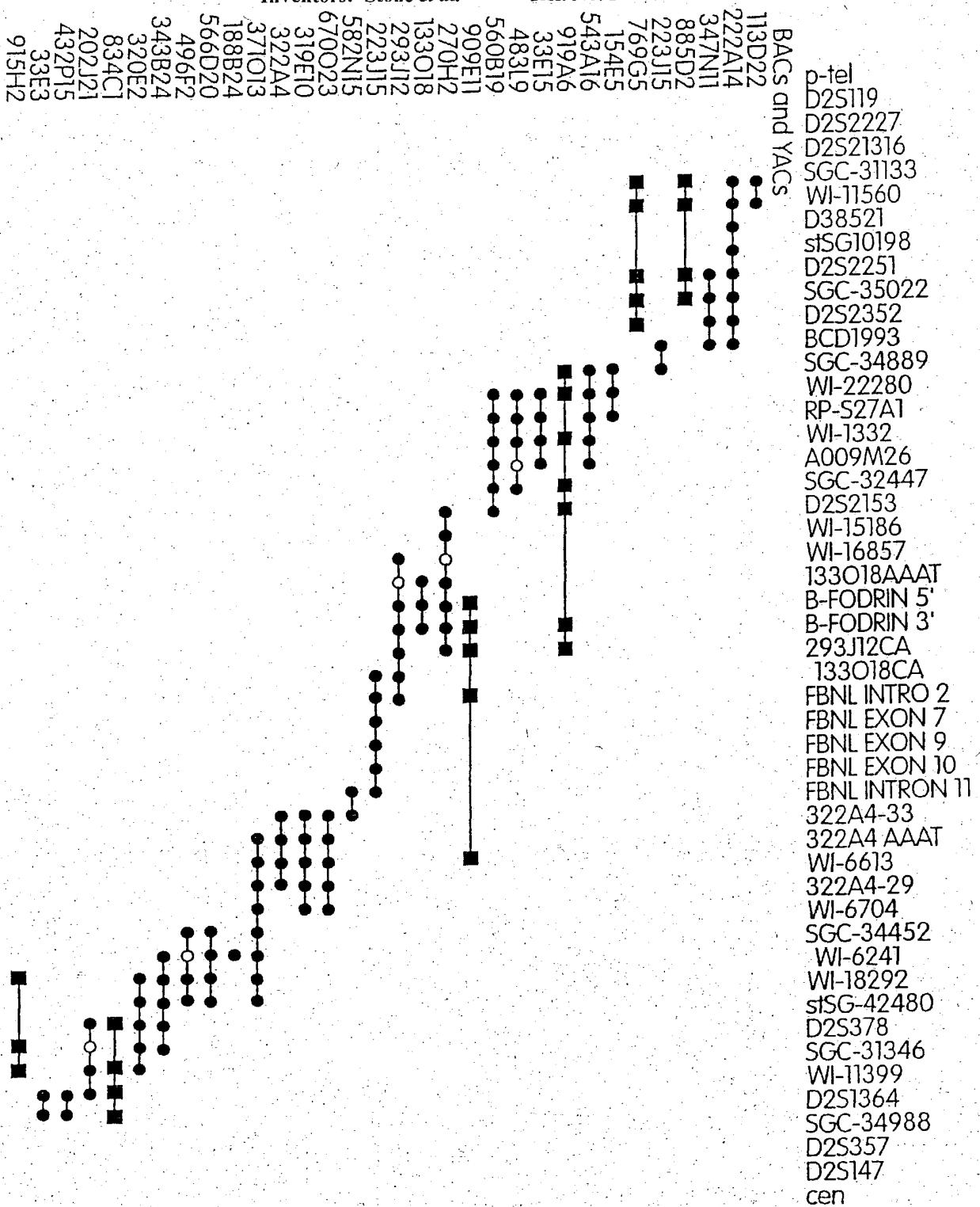
Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICSInventors: Stone *et al.* Ref. No. 21087.0019U3

FIG. 2B

SHEET 4 OF 12

Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS  
Inventors: Stone *et al.* Ref. No. 21087.0019U3

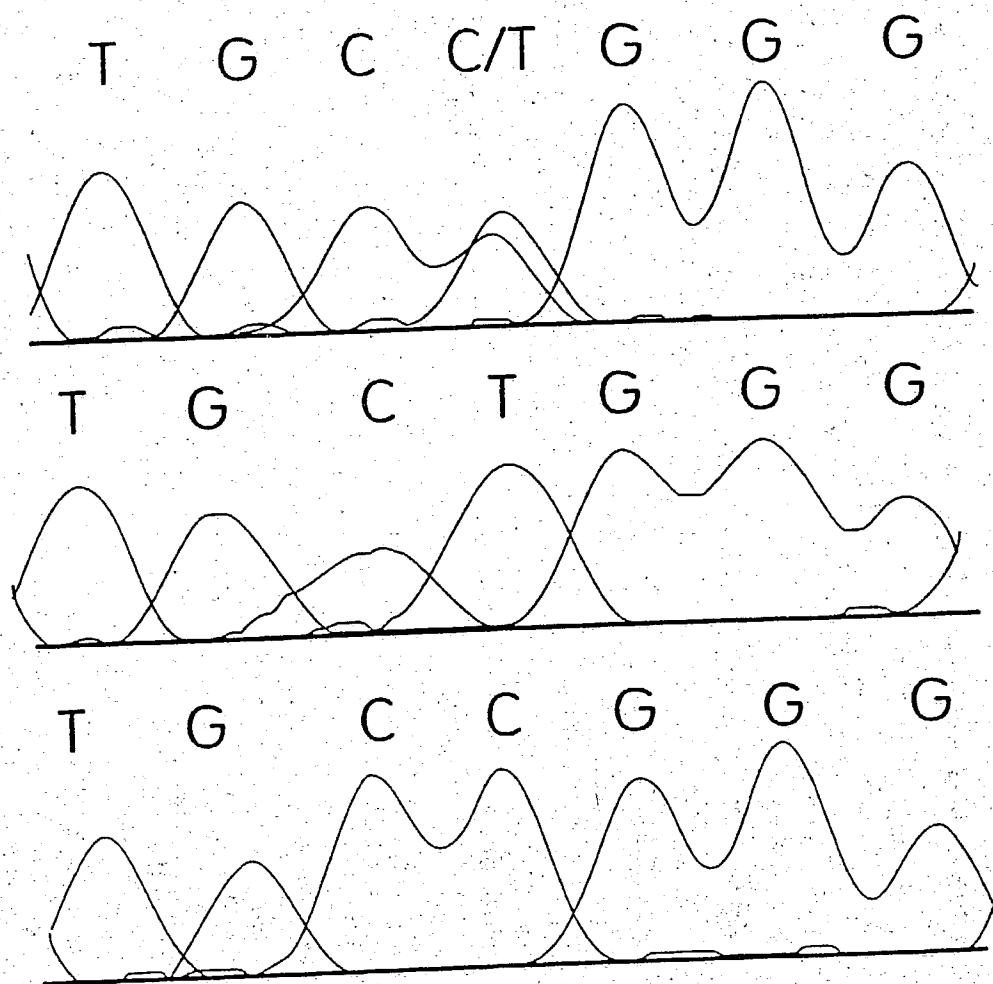
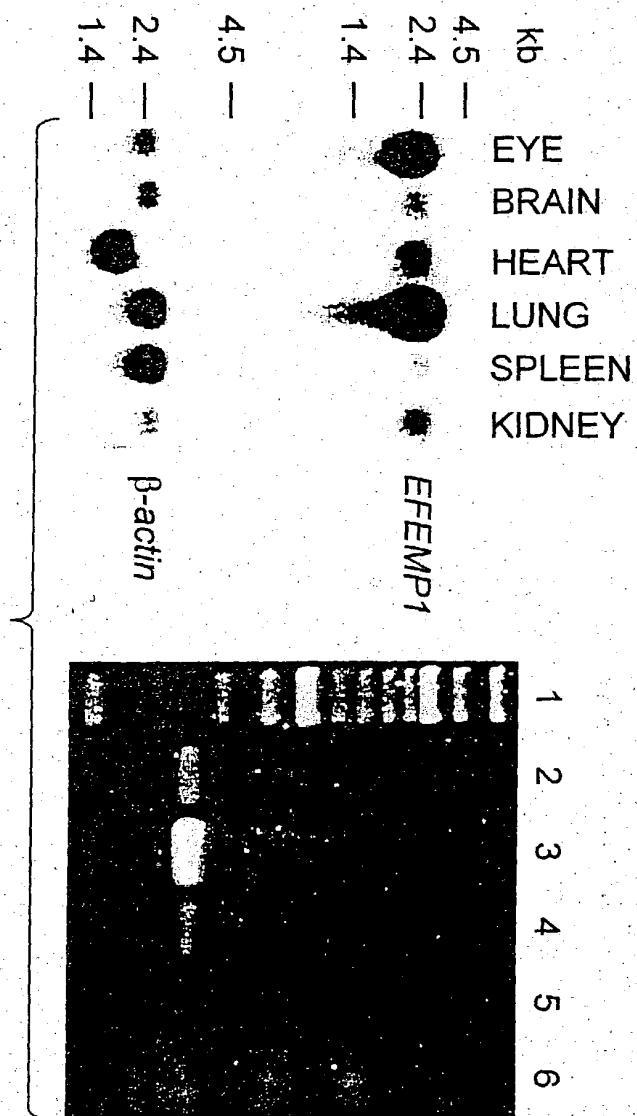


FIG. 3

SHEET 5 OF 12

Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS  
Inventors: Stone *et al.* Ref. No. 21087.0019U3



**SHEET 6 OF 12**  
**Title: MACULAR DEGENERATION**  
**DIAGNOSTICS AND THERAPEUTICS**  
**Inventors: Stone *et al.*** Ref. No. 21087.0019

Exon 1 Inventors: Stone *et al.* Ref. No. 21087.0019U3

Currently undetermined

### exon 2 partial transcript

AGTGTGCGAGATTTAAGCCGCACCTGGATTCCATAGGAGCTGGTTAGAACGCTGGGACGCTGAGCAGCT  
CCAGGGGACCGCCCGCTTAGCTTGCTTAAGAAAGGGGACCTCATCTCCCTGCCGGGCCAGGCCGCC  
CGCCCCGAAACTGGTACCTTGGGCTCGGGTGCATCCCTGGTTCGGTCCTAGGCAGCCTGAAACGAA  
GGTAGCGTGTGGGGACCCAGACTGTATAAGACAAAAGAGAACAGTCGCTTGGGCTGCCCTCCACA  
CAACCTGGGACTTTAAACAAAGCTGTGCGAGAGAAAGGCGTGGAAATGCCACTTGGAGAGTTGTG  
CTGGGGGATGTGAGAACGCTGTGAGACATGTGAGAAGGTCTAGTATTCTACTAGAACCTGGAGATTGCT  
CTCCGAGTTTGTATTGTAAAAAAATAAAAGCTTGAGGCCAAGGCAATTCATATTGGCT  
CACAGGTATTTTGTGTGCTGTGCAAGGAACCTGCTAGCTCAAG (SEQ ID No. 46)

### Complete intron 2

gtgagtatac tggaaagcc ttaacgttct agagaacaga gttttgcagcccccacctcca  
tcccccgaaa tggtggactg gggggcagga aatgcaccgtccccttgaa atggattact  
gttttttcct ttgcacccccc tctttctgcagcctgccttg taggtgcagt ataaaaatgc  
cgctgaatgt ctttgtatgtaaacagcgt agcaggatgg agtaacgtga aatgcaattc  
tacagcagttttacgtctt tgctgcctcg ttctggct accgagaagg ttcaggaggggaggggaga  
tgagaaagca gatttggaaagt tgagtatgtt ggtacgcctcagcctccca ccctcccttc  
ctcgcttgcgt ctcaactgcta aagttttgttacttcccccg cagcagatac taaacattag  
tttgcctgtt atttcttttag (SEQ ID No. 47)

### Exon 3

ATTCCACAAATG TTGAAAGCCC TTTTCCTAAC TATGCTGACT CTGGCGCTGG TCAAGTCACA  
GGACACCGAA GAAACCATCA CGTACACG (SEQ ID No. 48)

Start intron 3

gtaaagggtg atggaatttg aaataagttac ggttcagcgg gattctgtgacaaaaaaaaaaga ctttacacac  
ccctgcagt gtatttggc tatactctgtcgagggtat aaattaaca acacttcatt catgcttcat  
atctaagattcggttaaat tgcccccttg atccttcaa aagttcattg ggctcaccacctaagatagg  
aaccacatg taatcatttg tgcagggcta aaaatggatccgttcaaaa actaaaaacca aagaaggta  
catgttcca aaacattcaacaaattaatg ggtgtaaagg actggaaaac ctgactcct  
accacatgcagataaaacca atacgtcag aataagactc aagtcaagta agaacgttacccataaa  
gacacatggccttctgttatcatgacatgcattctcaagtaagtggccttattgttataaggctata  
tattcattctttgtataacttg ataattctaataaaaggcagacaacagttatgttaccaggatg  
catattggctaaagtggttaaaacgtaatgtgtgcaactccgtttgcatttctaattagcgtct  
ctgatattccaagtaatattgatttagttagttgcataggtgtacccatgttataaaaatattaaaaagat  
cacctgaccctcccactgctacaaatagttgtggtagaaac agagaaggacagtaactgacttcaattctggt  
gagttgttgcacctgttctgtttctgtttaatcagtgttaggcaaatgacattgtcctggatt  
ggaatatgaaaa gcacattttctactgctcccagttaaaattaagtaatcctactcgaaagaatgtgaaa  
aattttgaaaagaaaactctaaaaatgaactaattgtcaattactgtataataaac attatctcacttttgg  
taccacattatccctagaatgttagtattcatctggcaatgttccttcgttgcattccactataaat  
aaaatagcttaatacaatatttgttaact tcattgtttaatgcagttgtcctctgtcaagataaattaa  
gcaagaaaaatgaaggcatgtgtkttatctttaaatgaaaatgktttktattcagactaaacttactgccc  
ttctcangggagctaaaattactaccactttataatcatctcataaaagattttactctttcca  
gttgc (SEQ ID No. 49)

FIG.5A

SHEET 7 OF 12

**Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS**

End intron 3

Inventors: Stone *et al.* Ref. No. 21087.0019U3

Ref. No. 21087.0019U3

aaaatgcctg gtttcagaa ggggggttycc ataggctgtt tggtcagngggggngctaar gaggnnatt  
gaagctgggg actgttnggg ggaaccccncgttcttgc gagcttaant tanggagcng gtcagggggg  
gaaanggaggggcttaatn ctgtinanagg nttnaaaaaa aaaaaaaaaa ntccnggctggttnggggt  
ggggngggg gaaagggcc aaaaaaaaaa aaaaaatggtnnnnnnnn tttiacattt ccaatgtggg  
aaaaaaggca aattaataaagagcagtca agaagttgga gaagattagt ctcaaaacag  
aaaagaagatggactggc anctgtacca aaaagaacag aagagtttag gcagctgtatggttgagaatg  
gaccggcga gctgtccat gcacagactt gtctttgaaaaaaaaagcga tagaatgtta aaccaccccat  
ctcatcatat atcttaggacttttagcacaag gattgttgcc ataagaatga agcttttaga  
gtgattttcttagggaatgga cacaccaatt aactgtctcc tggccccacc tttgatttttcttcacag  
(SEQ ID No. 50)

### Exon 4

CAATGCACTG ACGGATATGA GTGGGATCCT GTGAGACAGC AATGCAAAG (SEQ ID No. 51)

complete intron 4

gtgagccagcttcaaaggacttcattgtatgccttgtccatctttgtcactttccctgt  
ctgtttataccaaaaaggcatgacattatattacatgtttgatcccttagaaarattcctgacttat  
tttattactqaccacaq (SEQ ID No. 52)

### Exon 5

ATATTGATGA ATGTGACATT GTCCCAGACG CTTGTAAAGG TGGAATGAAG TGTGTCAACC  
 ACTATGGAGG ATACCTCTGC CTTCGAAAA CAGCCCAGAT TATTGTCAAT AATGAACAGC  
 CTCAGCAGGA AACACAACCA GCAGAAGGAA CCTCAGGGC AACCACCGGG GTTGTAGCTG  
 CCAGCAGCAT GGCAACCAGT GGAGTGTTCG CCGGGGGTGG TTTTGTGGCC AGTGCCTGCTG  
 CAGTCGCAGG CCTGTAAATG CAGACTGGCC GAAATAACTT TGTCTATCCGG CGGAACCCAG  
 CTGACCTCA GCGCATTCCC TCCAACCCCTT CCCACCGTAT CCAGTGTGCA GCAGGCTACG  
 AGCAAAGTGA ACACAACGTG TGCCAAG (SEQ ID No. 53)

Start intron 5

Gtaggaaaacttatttcctaacccttgaaagctgcaggagaacccatgaagcttgaattcgatcca  
cgttq (SEQ ID No. 54)

End intron 5

Tttggcattctgaaatttgcaatgaaagattatacawacatgtgccctgctttggaaaaacaaacagc  
tacmggraacggataatattaaaggtagtgcacccagttatggtagatttttagaaatttgtcaatgg  
aattatctcaaatacaatataattggatggaaaaggcaagtatcatacaatctattaaaattttacataca  
aataccatatgttctaattggatgcattccctgtctaacaaaagtacaaaacatctcagggaaggattcatt  
ccgagacagtggtagctgatgggtcaaggatgaggatggtagctgttagctgtatctgaaaatgttctta  
aaaaacaaaatgagccaagaccaacatgacaaaatgttagcattttaaatctgagcagactcactggatt  
gaaaaattatttctgaacactgaaataatttataattttaaacatttcaatgcaagaacattataaacttt  
aagaataaagtwwaaaatttagctttagaaagtggcmaatggargaaatatacatcttcacaactgacaatyyt  
tytstgtctwgayatgtctswgacag (SEQ ID No. 55)

## Exon 6

ACATAGACGA GTGCACTGCA GGGACGCACA ACTGTAGAGC AGACCAAGTG TGCATCAATT  
TACGGGGATC CTTTGCATGT CAGTGCCTTC CTGGATATCA GAAGCGAGGG GAGCAGTGC TAG  
(SEQ ID No. 56)

FIG. 5B

SHEET 8 OF 12

# Title: MACULAR DEGENERATION DIAGNOSTICS AND THERAPEUTICS

Start intron 6

**Inventors:** Stone *et al.*      **Ref. No. 21087.0019U3**

End intron 6

## Exon 7

ACATAGATGA ATGTACCATC CCTCCATATT GCCACCAAAG ATGCGTGAAT ACACCAAGGCT  
CATTTATTG CCAGTGCAGT CCTGGGTTTC AATTGGCAGC AAACAACAT ACCTGCGTAG  
(SEQ ID No. 59)

Start intron 7

Gtaaggccctttgagaacttgtctgttttcacaaaaggaaacccatgccatgtgcagtatttccagt  
tttctatgttcttgagtaaatagttccatcgacttcccttcagcaatcataaagytgcaggagaacccatga  
agcttgaattcgatccacgtgg (SEQ ID No. 60)

End intron 7

aaactttccactggaaaagtgcattctgattttacatgccttttgccttcagaaatgcatacggt  
: cacaacaattaaaagattggcatgacatggaaaaaa:tctagtgtggaaaatgcctttcaacaatatt  
t:cagtgcatttagaagcattgcaaaactccgtatgggtctcaaaaggcttatgttataattgtatggaaattta  
acagaacccattnaaaaagttataaa:atagccacagataaatctccagttaccagcattgcctgaagaagac  
catatccagtataagttgttataawcaattattttatagaaattggcatttgtwtcttgaaaccaacaaaagaa  
aaatccgaat:mccggaaktgttatatttttagaagcattaaattcccttgganagattnatcacacatcnna  
ctaactgtcattcctagaaaaatattcgttattccnnaagaagtatatgacagacgttgttagttgttccc  
acaaatatganaccnnaatggatgttctccagtgagctctgcagggcaaataattcagctaggaaattactca  
cttgtcagcagatgacgttaggtacaaaagagaaggatgttataagtstaymtatmtgtgtgtatay  
tatacatatacaymwmymtatayatamratttttcwag (SEQ ID No. 61)

## Exon 8

ATATAAATGA ATGTGATGCC AGCAATCAAT GTGCTCAGCA GTGCTACAAAC ATTCTGGTT  
CATTCATCTG TCAGTGCAAT CAAGGATATG AGCTAAGCAG TGACAGGCTC AACTGTGAAG  
(SEQ ID No. 62)

FIG.5C

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**Title: MACULAR DEGENERATION**

## DIAGNOSTICS AND THERAPEUTICS

Inventors: Stone *et al.* Ref. No. 21087.0019U3

Start intron 8

Ref. No. 21087.0019U3

End intron 8

Aaaaaaatatatatgtgtgtgtgtgtgtgtgttatattaaacccagncaacttaaaaatgt  
gccaagtcacacagtcgcaggaataggacaanaagccagatcttttatataataggtta,gatataattt  
ttcctccttanaatataaataa,tttaatttatataattttatataatagatattttaaatcttataattt  
atatatatataatttatataatataatccaaagttagtgtgcacaaactttcaactctgtgtcctt  
tctcttgtctaattcaacag (SEQ ID No. 64)

### Exon 9

ACATTGATGA ATGCAGAAC C TCAAGCTACC TGTGTCAATA TCAATGTGTC AATGAACTG GGAAATTCTC  
ATGTATGTGC CCCCAGGGAT ACCAAGTGGT GAGAAGTAGA ACATGTCAAG  
(SEQ ID No. 65)

Start intron 9

gtaagtttattttttttcatatgttagtgtttagtttagcaggaaagagacaagaggaagttaggatt  
ctccatatagacttcattttccacttcaatatacaatattaaagctntttccctgtcatcataaaaata  
tatacatctcataaagaggggattctatgtaangccacnntttcgctctaaaagataataattttata  
aatatttgatatgtattctatgtaacctacatcatctnttgagatacatctcaaataatccactgaaaaaga  
ttcagttattaaaangttcacctgtgagttgagttanagcataagctcaatatggagttaaacatacctc  
catccagtttagccctctaaaacncanggattataaattgcgtaaaaatgttaggtgctaaaaaagtccagct  
aatatgtgtaaaatatagttgaaatattttagagaaaactactagccccaaaatagctaatacgaccctgggtcca  
gtttcaaaataaacattcagatcttccacccatatacgtaaagkggaaagaggcagctccccacaatggatga  
tttcagagttctcaggaagatctaaaaaaaaaaaaggaccctacccatgttgcattgttagttaaaaattttc  
ttaacaggggaaaggactgtcanataaaacccaaaacgtaaaaatccctggaaaagctagtncaaacncttaaat  
ttacncaaagcaccaaaagaataaaaatgaccaancctgacanaaaacctgtttgaatcccagctccactgt  
nttcagtcgcnaatnttgaacaaaattatcaaactactntgagcctcagntccctcatttgaaaagggagtt  
ggggaaatttagggaaatancntaaaaatanttgtaaactataaagctgtncaggtcaaggggaaaaatttt  
atnaaatttac (SEQ ID No. 66)

FIG.5D

SHEET 10 OF 12

**Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS**

Inventors: Stone *et al.* Ref. No. 21087.0019U3

End of intron 9

AgcctcttcctaacttcttttccttacagtccaaaattgttatgcttatgaggtggAACACTTCATA  
gtttcacttcgtgtgtgcctcgtggacagtataatccactcccagcatgttcagttactgaaaccagg  
atttctagccttaccttctccaagttctgaaagagatgataagtcgcctccatagttatgttcctgaa  
tttctcagcttgaaagccttcgtccccagccatgaaaactccatctaaccaccaccctcaaggcacgtt  
gagatgccttccttccttcgtccatcccggcaaaattaccaactctgtccacatccccca  
gtatacttatctatcttacttaattccatttactttctaatgttatcatatacacattccctcaattataat  
gtccctgtatgacaagaactgggtttactttatataaggcagagtcatgttgtaacattggggtttgaattca  
acagatgaacaataggtgttataaaaatcatgaaatgacacatattaatctgcctagaatgtctcagctt  
gtctgtcctgaattcaatacatgaaacacccagtcgtgtctaaaaggcaggttgaacacagtcagatgtct  
cacaccccttccttgcaaacagaatctgcctgttatgatttaatttagatcagttcatttagtttagtagta  
aactcttgaccctacatctacag (SEQ ID No. 67)

## Exon 10

ATATAATGA GTGTGAGACC ACAAAATGAAT GCGGGGAGGA TGAAATGTGT TGGAAATTATC  
ATGGCGGCTT CCGTTGTTAT CCACGAAATC CTTGTCAAGA TCCCTACATT CTAACACCAAG AGAA  
(SEQ ID No. 68)

## intron 10

gtaagaaaaa tcagaacttt tgaaaagttag gatTTCTGG tcttaccaagccaaactgct gaatactttt  
gtttgtctct gcag  
(SEQ ID No. 69)

### Exon 11

CCGATGTGTT TGCCCAGTCT CAAATGCCAT GTGCCGAGAA CTGCCCCAGT CAATAGTCTA  
CAAATACATG AGCATCCGAT CTGATAGGTC TGTGCCATCA GACATCTTCC AGATACAGGC  
CACAACATT TATGCCAACAA CCATCAATAC TTTTCGGATT AAATCTGGAA ATGAAAATGG  
AGAGTTCTAC CTACGA (SEQ ID No. 70)

Start intron 11

gtaaatcc tgaaggcagc cttactatt gagaagatg ggagttgttgggttgg ttgttgttgg  
tggtgttgg tatccacatg tggaaaggaaagcaacattt aaaagtgtcttnatgttaggcattgttaag  
gccttcacgttacattttcatttatcccttggtaacactgcacatagatataatattcatctccattt  
ttacagaggagaaaagttagatgcagaaagatgttagcatccctgaaatcactcaaataattaaagtggcaga  
ctctgatagatgtgtgaccacgaaaataacaagctccatcccccgctctgccccacccaacataacc  
cccaagttagtatactaatcattgatggtaattaattatacatagacatacatataattcaaaacccaaaataa  
ttccctggagctctaaagagttttcagacatcatgaattcatcattgttacattcacaagacagttgttca  
caccgaactaaaacctataagtatgtgagaagtgcacccacccctcccgacagtatgtgtcaagttagtgttacc  
ttcttgccaacttctggctggcagttatggagtcatctccatctttcattgcctgtgttaatctactttctg  
aattctgccatcccttccactgtctctgggttatcttgcctccatccctatctctctccatata  
aactggctccgtactccatgtccctcagttggcttgcacatccctgacccaaagcctcagccct  
ccagttccctacaaaatctcagcaagttcatttaaaggtaaaatttggacatatttaataacggctcaccac  
ttcatgtaaaaatgtggcacccatccaagcagttgcagagttaccggtaactgtttcatgtaatgtatgttay  
tcatcccaatgg SEQ ID No. 71)

End intron 11

Tcccttttttttctaaaaaggnaaccnatggcccaagntgnaaaaanaaaaaggccncttgnttccaggttaaaaattccnatttcccctwaagtttagktttggaaaggccccacttcncannaaaaggaaaaatgnntacmaanaggggggattcaaaacnaaaaactttttaaaaaaaaaaaaaaggcaagtcttgaaacttggagctaattgactgtattagacaaggataagagccaagaagagttgaaccagaagaaggaccagaagttggctctttataccaccttcaaaattctcccttaattcttataggaggtataactaacaaagcatagaaactccaatccaagaaaattattcttccttctctatttctttattttag (SEQ ID No. 72)

SHEET 11 OF 12

Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS  
Inventors: Stone *et al.* Ref. No. 21087.0019U3

Exon 12

CAAACAAGTC CTGTAAGTGC AATGCTTGTG CTCGTGAAGT CATTATCAGG ACCAAGAGAA  
CATATCGTGG ACCTGGAGAT GCTGACAGTC AGCAGTATAAG GGACCTCCG CACAAGCTCT  
GTGTTAACAT AGTGGGGCCA TTTTCATTT AG (SEQ ID No. 73)

3' utr

tcttttctaa gagtcaccca caggcattta agtcagccaa agaatattgt taccttaaag  
caactatTTTA tttatagata tatctagtgc atctacatct ctatactgta cactcaccca  
taacaaacaa ttacaccatg gtataaagtg ggcatttaat atgtaaagat tcaaagttt  
tcttttattac tatatgtaaattagacatta atccactaaa ctggcttct tcaagagac  
taagtataca ctatctggtg aaacttggat tcTTTcctat aaaagtggga ccaagcaatg  
atgatcttct gtggtgctta agggaaactta ctagagctcc actaacagtc tcataaggag  
gcagccatca taaccattga atagcatgca agggtaagaatgagttttta actgtttgt  
aagaaaaatgg aaaaggctaa taaagatatatTTCTTtaga aaatggggat ctgccatatt  
tgtgttggtt ttatTTTcatatccagcct aaagggtggtt gtttattata tagtaataaa  
tcattgtgtacaacatgtc ggTTTCTGta gggTattttt aattttgtca gaaatttttagattgtgaata  
ttttgtaaaa aacagtaagc aaaattttcc agaattccaaatgaacca gataccccct agaaaattat  
actattgaga aatctatggggaggatatga gaaaataaaat tcTTTctaaa ccacattgg  
actgacctgagaagcaac tcggaaaata taataacatc cctgaattca ggcattcacaagatgcagaa  
caaaatggat aaaaggatatt tcactggaga agttttaaTTTctaagtaaa atttaaatcc taacacttca  
ctaatttata actaaaatTTctcatcttcg tacttgatgc tcacagagga agaaaatgtat  
gatggTTTTTattccctggca tccagagtga cagtgaactt aagcaaattt ccctcctacccaattctatg  
gaatattttta tacgtctcct tggTTTaaat ctgactgctttactttgatg tatcatattt ttaaataaaaa  
ataaaatattc ctttagaagatcactctaaaa  
(SEQ ID No. 74)

FIG.5F

SHEET 12 OF 12

Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS  
Inventors: Stone *et al.* Ref. No. 21087.0019U3

M LKALFLTMLTL ALVKSQDTE ETITYTQCTD GYEWPVRQQ  
CKDIDECDIV PDACKGGMKC VNHYGGYLCL PKTAQIIVNN EQPQQETQPA  
EGTSGATTGV VAASSMATSG VLPGGGFVAS AAAVAGPEMQ TGRNNFVIRR  
NPADPQRIPS NPSHRIQCAA GYEQSEHNVC QDIDECTAGT HNCRADQVCI  
NLRGSFACQC PPGYQKRGEQ CVDIDECTIP PYCHQRCVNT PGSFYCQCSP  
GFQLAANNYT CVDINECDAS NQCAQQCYNL LGSFICQCNO GYELSSDRLN  
CEDIDECRTS SYLCQYQCVN EPGKFSCMCP QGYQVVRSSRT CQDINECETT  
NECREDEMCW NYHGGFRFCYP RNPCQDPYIL TPENRCVCPV SNAMCRELPQ  
SIVYKYMSIR SDRSVPSDIF QIQATTIYAN TINTFRIKSG NENGEFYLRQ  
TSPVSAMLVL VKSLSGPREH IVDLEMLTVS SIGTFRTSSV LRLTIIVGPF  
SF (SEQ ID NO.: 2)

FIG. 6